ASNS gene

asparagine synthetase (glutamine-hydrolyzing)

Normal Function

The *ASNS* gene provides instructions for making an enzyme called asparagine synthetase. This enzyme is found in cells throughout the body, where it converts the protein building block (amino acid) aspartic acid to the amino acid asparagine. Another amino acid called glutamine helps in the conversion and is itself converted to the amino acid glutamic acid during the process. It is thought that asparagine synthetase helps to maintain the normal balance of these four amino acids in the body.

Asparagine is needed to produce many proteins but also plays other roles. Asparagine helps to break down toxic ammonia within cells, is important for protein modification, and is needed for making a certain molecule that transmits signals in the brain (a neurotransmitter).

Although asparagine can be obtained through the diet, the amino acid cannot cross the protective barrier that allows only certain substances to pass between blood vessels and the brain (the blood-brain barrier). As a result, brain cells rely solely on asparagine synthetase to produce asparagine.

Health Conditions Related to Genetic Changes

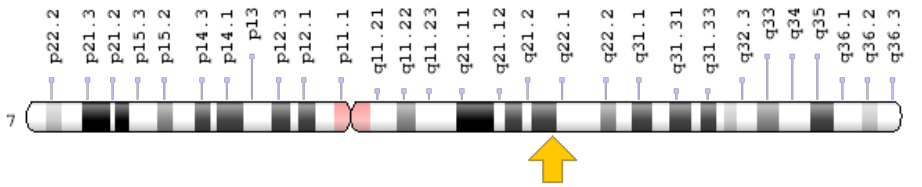
Asparagine synthetase deficiency

At least 15 mutations in the *ASNS* gene have been found to cause asparagine synthetase deficiency, a severe condition that causes neurological problems soon after birth. In most cases, these mutations replace single amino acids in the enzyme. The altered enzymes have little or no function. Asparagine from the diet likely makes up for the enzyme's inability to produce the amino acid in cells. However, because asparagine cannot cross the blood-brain barrier, brain cells in people with asparagine synthetase deficiency have a shortage (deficiency) of this amino acid. The exact effect of asparagine synthetase deficiency on brain cells is unknown, but because of the severe features of this condition, it is clear that asparagine is necessary for normal brain development. A lack of asparagine in developing brain cells leads to poor brain development and the severe neurological problems in individuals with asparagine synthetase deficiency. It is unclear whether a lack of glutamic acid or a buildup of glutamine impacts the signs and symptoms of this condition.

Chromosomal Location

Cytogenetic Location: 7q21.3, which is the long (q) arm of chromosome 7 at position 21.3

Molecular Location: base pairs 97,851,677 to 97,928,441 on chromosome 7 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- aspartate ammonia ligase
- glutamine-dependent asparagine synthetase
- TS11
- TS11 cell cycle control protein

Additional Information & Resources

Educational Resources

- Biochemistry (fifth edition, 2002): An Adenylated Intermediate Is Required to Form Asparagine from Aspartate
  https://www.ncbi.nlm.nih.gov/books/NBK22459/#A3353

Clinical Information from GeneReviews

- Asparagine Synthetase Deficiency
  https://www.ncbi.nlm.nih.gov/books/NBK525916

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ASNS%5BTIAB%5D%29+OR+%28asparagine+synthetase%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- ASPARAGINE SYNTHETASE
  http://omim.org/entry/108370

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/ASNSID44323ch7q21.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=ASNS%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:753
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:440
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/440
- UniProt
  https://www.uniprot.org/uniprot/P08243

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